



Schinzal-Giedion syndrome

Schinzal-Giedion syndrome is a severe condition that is apparent at birth and affects many body systems. Signs and symptoms of this condition include distinctive facial features, neurological problems, and organ and bone abnormalities. Because of their serious health problems, most affected individuals do not survive past childhood.

Children with Schinzal-Giedion syndrome can have a variety of distinctive features. In most affected individuals, the middle of the face looks as though it has been drawn inward (midface retraction). Other facial features include a large or bulging forehead; wide-set eyes (ocular hypertelorism); a short, upturned nose; and a wide mouth with a large tongue (macroglossia). Affected individuals can have other distinctive features, including larger than normal gaps between the bones of the skull in infants (fontanelles), a short neck, ear malformations, an inability to secrete tears (alacrima), and excessive hairiness (hypertrichosis). Hypertrichosis often disappears in infancy.

Children with Schinzal-Giedion syndrome have severe developmental delay. Other neurological problems can include severe feeding problems, seizures, or visual or hearing impairment.

Affected individuals can also have abnormalities of organs such as the heart, kidneys, or genitals. Heart defects include problems with the heart valves, which control blood flow in the heart; the chambers of the heart that pump blood to the body (ventricles); or the dividing wall between the sides of the heart (the septum). Most children with Schinzal-Giedion syndrome have accumulation of urine in the kidneys (hydronephrosis), which can occur in one or both kidneys. Affected individuals can have genital abnormalities such as underdevelopment (hypoplasia) of the genitals. Affected boys may have the opening of the urethra on the underside of the penis (hypospadias).

Bone abnormalities are common in people with Schinzal-Giedion syndrome. The bones at the base of the skull are often abnormally hard or thick (sclerotic), or the joint between the bones at the base of the skull (occipital synchondrosis) can be abnormally wide. In addition, affected individuals may have broad ribs, abnormal collarbones (clavicles), or shortened bones at the ends of the fingers (hypoplastic distal phalanges).

Children with this condition who survive past infancy have a higher than normal risk of developing certain types of tumors called neuroepithelial tumors.

Frequency

Schinzal-Giedion syndrome is very rare, although the exact prevalence is unknown.

Genetic Changes

Schinzel-Giedion syndrome is caused by mutations in the *SETBP1* gene. This gene provides instructions for making a protein called SET binding protein 1 (SETBP1), which is known to attach (bind) to another protein called SET. However, the function of the SETBP1 protein and the effect of its binding to the SET protein are unknown.

The *SETBP1* gene mutations that have been identified in Schinzel-Giedion syndrome cluster in one region of the gene known as exon 4. However, the effects of the mutations on the function of the gene or the protein are unknown. Researchers are working to understand how mutations in the *SETBP1* gene cause the signs and symptoms of Schinzel-Giedion syndrome.

Inheritance Pattern

Schinzel-Giedion syndrome results from new mutations in the *SETBP1* gene and occurs in people with no history of the disorder in their family. One copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Schinzel-Giedion midface retraction syndrome
- Schinzel Giedion syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Schinzel-Giedion syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1849294/>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Birth Defects
<https://medlineplus.gov/birthdefects.html>

Genetic and Rare Diseases Information Center

- Schinzel Giedion syndrome
<https://rarediseases.info.nih.gov/diseases/117/schinzel-giedion-syndrome>

Educational Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology: Schinzel-Giedion Midface Retraction Syndrome
<http://atlasgeneticsoncology.org/Kprones/SchinzelGiedionID10129.html>
- Boston Children's Hospital: Hydronephrosis
<http://www.childrenshospital.org/conditions-and-treatments/conditions/hydronephrosis>
- Disease InfoSearch: Schinzel Giedion syndrome
<http://www.diseaseinfosearch.org/Schinzel+Giedion+syndrome/6438>
- MalaCards: schinzel giedion syndrome
http://www.malacards.org/card/schinzel_giedion_syndrome
- My Child Without Limits: Developmental Delay
<http://www.mychildwithoutlimits.org/?page=developmental-delay>
- Orphanet: Schinzel-Giedion syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=798
- University of Michigan Health System: Developmental Delay
<http://med.umich.edu/yourchild/topics/devdel.htm>

Patient Support and Advocacy Resources

- My Child Without Limits
<http://www.mychildwithoutlimits.org/?page=home>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/schinzel-giedion-syndrome/>
- University of Michigan Health System: Developmental Delay
<http://med.umich.edu/yourchild/topics/devdel.htm>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28schinzel-giedion+syndrome%5BTIAB%5D%29+OR+%28schinzel+giedion+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- SCHINZEL-GIEDION MIDFACE RETRACTION SYNDROME
<http://omim.org/entry/269150>

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<http://atlasgeneticsoncology.org/Kprones/SchinzelGiedionID10129.html>
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Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21371013>

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